2018

PRESS KIT

Clinic *for* Special Children

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Our Mission

To serve children and adults who suffer from genetic and other complex medical disorders by providing COMPREHENSIVE MEDICAL, laboratory, and consultative services, and by INCREASING AND DISSEMINATING KNOWLEDGE of SCIENCE AND MEDICINE.

Our History

Internationally recognized as an innovative MEDICAL & SCIENTIFIC organization

In order to provide local, affordable services Dr. Morton and his wife Caroline partnered with the local Amish and Mennonite populations to found the non-profit Clinic for Special Children in 1989. In 1990, the original post-and-beam style building was raised largely by volunteers from the Plain communities. In 2000, an expansion was added to the original building, adding more office and exam room space.

Since its founding, the Clinic for Special Children has endeavored to integrate advanced laboratory techniques into every day primary pediatric care. A molecular geneticist, Dr. Erik Puffenberger, was hired in 1998, opening the door for faster, more accurate diagnoses using molecular techniques. Today, the Clinic operates a CLIA certified, PA state licensed clinical laboratory focusing on biochemical and genetic tests that are most important to our patient population's needs.

The integration of clinical and laboratory services has generated many opportunities for research and scholarly work. This includes natural history studies, clinical trials, and gene variant identification. Research and clinical efforts were enhanced by hiring a second pediatrician in 2001, the Harvard trained Dr. Kevin Strauss. Dr. Strauss became Medical Director in 2008 and has authored many of the seminal works in the treatment of disorders observed at CSC. Collaborations with academic and industry partners has helped to bring resources to determining the molecular consequences of the variants identified in the Clinic's lab.

Although the Clinic was founded to treat metabolic disorders, the organization now treats patients with genetic disorders of all types. The organization is envisioned as a medical home for patients, meaning they can see physicians knowledgeable about their disorder when they are sick and when they are well. In order to enhance the idea of a medical home, the Clinic has forged collaborative relationships with individual physicians and healthcare organization to provide affordable specialty care on site, making the services more accessible to patients.

After many years of distinguished service, Caroline Morton retired from her staff position and Dr. Morton departed the organization in 2016. However, their legacy of tireless service lives on in the day to day service provided by the Clinic.

Internationally recognized as an innovative medical and scientific organization, the Clinic for Special Children has remained on the cutting edge of genomic and translational medicine. However, despite the innovative and pioneering nature of the organization, the Clinic has remained focused on its most important task: providing care to vulnerable individuals with genetic disease.

2017 Quick Facts

3,914 *biochemical & genetic tests*

1,052 *active patients*

17 *staff members*

1,606 *patient visits*

42 *states*

Manage 264 *known variants that cause disease*

5 *peer- reviewed publications*

10 *family days*

17 *countries*

39 *NEW disease-causing variants*

Key Staff Bios

KARLLA W. BRIGATTI, MS, LCGC

Research Operations Director

Karlla W. Brigatti, MS LCGC, joined the Clinic for Special Children as its first genetic counselor in 2014. She earned her Bachelor of Science in Cell and Molecular Biology from the University of Pittsburgh in 1994 magna cum laude and her Master of Science In Human Genetics from Sarah Lawrence College in 1998.

Prior to joining the Clinic, she was the senior coordinator of the FASTER Trial at Columbia University, the largest NIH-funded trial in Obstetrics and Gynecology to date, and the founding coordinator for the Center for Prenatal Pediatrics at Columbia University, introducing multidisciplinary and state-of-the-art innovation to the care of highly complex pregnancies before and after delivery. After moving to the Lancaster area in 2006, she served as Senior Genetic Counselor in Clinical Genetics, Pediatric Oncology, and Neurology at the Children's Hospital of Philadelphia (CHOP), working with families from across the globe in the Friedreich Ataxia Center of Excellence at CHOP on various natural history and clinical drug trials for the condition.

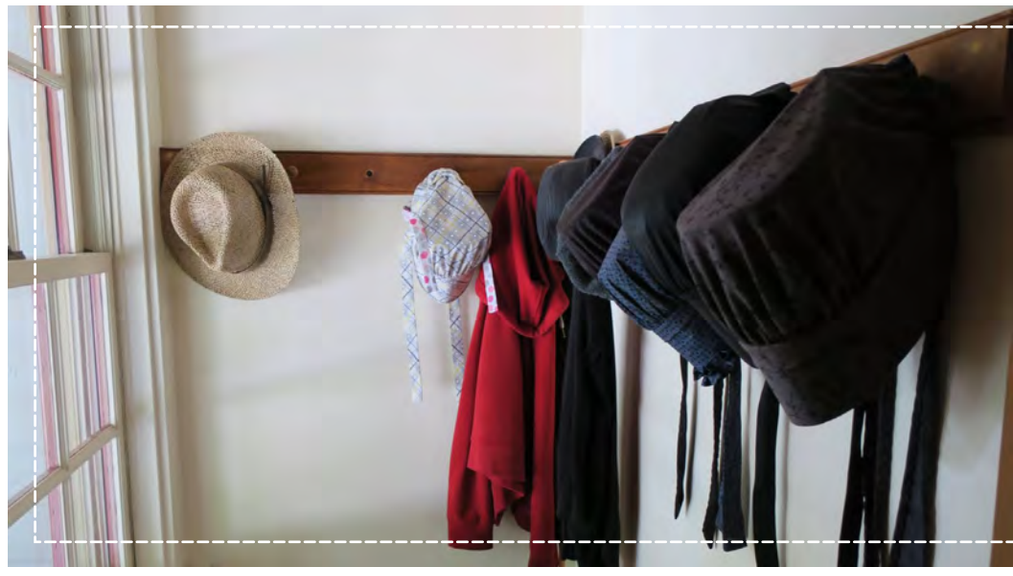
She has authored over 25 lay and scientific publications, mentored undergraduate, graduate, and medical students, and served on the Human Genetics Faculty at Sarah Lawrence College and the Human Genetics and Genetic Counseling program at Jefferson College of Biomedical Sciences. She recently completed a one-year program in Rare Disease Clinical Research Training through the National Institutes of Health. In addition, she serves on the Board of the CROWN Foundation, promoting research in women's and newborn health. Her research interests include gene discovery, implementation of personalized medicine, and rare disease advocacy. She is certified by the American Board of Medical Genetics and is a member of the National Society of Genetic Counselors.

VINCE CARSON, MD

Pedatric Neurologist

Dr. Vincent Carson earned a Bachelor of Science degree in Biochemistry and Molecular Biology from Penn State University in 2004 and a Medical Degree from Drexel University in 2011. He completed his pediatric and child neurology training at the Children's Hospital of Pittsburgh. He has affiliations with the American Academy of Neurology and the Child Neurology Society.

Dr. Carson grew up in Lancaster County and followed the work of the Clinic for many years before joining the staff in 2016. Dr. Carson committed to working for the Clinic as he was heading into his final year of residence at the Children's Hospital of Pittsburgh. He has a special interest in the genetics of brain disease. As a neurologist he specializes in disorders of the brain, spinal cord, nerves, and muscles. His responsibilities at the Clinic include providing care for patients, serving on the organization's on-call rotation, reading EEGs, and participating in research protocols.



ADAM HEAPS, MS, MBA

Executive Director

Adam earned a Bachelor of Science degree in biology from Franklin & Marshall College in 2008, a Master of Science degree in biology from Millersville University in 2014, and a Master of Business Administration degree from Saint Joseph's University in 2018. The thesis for is MS in Biology degree focused on using next generation sequencing technology to characterize known genetic variants in CSC's patient population.

In 2010, Adam joined the Clinic for Special Children's staff as a Laboratory Technician. He was responsible for running clinical and research assays including amino acid quantitation by HPLC, urine organic acids by GC/MS, and DNA isolation from whole blood. In 2012 Adam was promoted to Laboratory Scientist, a position he still holds. In this position, he was responsible for Sanger sequencing, genotyping variants with probe based assays, and coordinating research samples. He has co-authored three peer reviewed papers with other CSC staff members.

In 2014, Adam was appointed the Administrative Director by CSC's board of directors and in 2016 he was named Executive Director. He is responsible for financial management, strategic planning, collaborative relationships, facilities, and human resources. As the Executive Director, Adam is part of the clinic's five-person leadership team. He still occasionally does lab work and continues to serve on the laboratory on-call schedule.

"I see a very dedicated staff who day in & day out is thinking about how you can work with children with difficult circumstances and give them the best shot at life."

- Adam Heaps, MS, MBA, Executive Director

ERIK G. PUFFENBERGER, PHD

Laboratory Director

Dr. Puffenberger received a B.A. in biology from Swarthmore College in 1987, and a Ph.D. in human genetics from Case Western Reserve University in 1996.

In 1988, he accepted a position with Dr. Victor McKusick as an editorial assistant for Mendelian Inheritance in Man, a catalogue of human genetic diseases and traits. That gap year at Johns Hopkins Hospital turned into a four-year experience which also included genetic research in the laboratory of Dr. Claire Francomano. One of the projects he worked on during this time was the first description of fibrillin gene mutations as a cause of Marfan syndrome. The research of Drs. McKusick and Francomano focused significantly on the Amish of Lancaster County. As such, Erik spent many hours compiling Amish pedigrees for these studies, and frequently accompanied the researchers on field trips to Pennsylvania and Ohio. These experiences helped cultivate his interest in founder populations.

Erik enrolled in graduate school in 1991 at the University of Pittsburgh to study human genetics with Dr. Aravinda Chakravarti. As Erik began work in the laboratory, he discovered that a sizable collection of Hirschsprung disease families in the Chakravarti study were Mennonites from Lancaster County. Eventually, thirty-two families were used to genetically map and identify the cause of Hirschsprung disease in these families. During these studies, Dr. Chakravarti moved his laboratory to Case Western Reserve University in Cleveland, OH, where Erik received his Ph.D. in 1996.

Following graduation, Erik did a brief post-doctoral fellowship with Dr. Chakravarti. It was during this fellowship that he became acquainted with Dr. D. Holmes Morton and the Clinic for Special Children. The clinic offered him a position as laboratory scientist and he began work in January 1998. In 2000, he assumed the role of Laboratory Director. Today, his work involves implementation of molecular techniques for routine diagnosis, research into the genetics of isolated populations, development of molecular strategies for newborn screening, and identification of novel disease genes by genetic mapping and exome sequencing.

TERESA RINEER

Development Director

Teresa graduated magna cum laude from Millersville University with her Bachelors of Science degree in communications and a concentration in public relations. She is currently a member of the Association of Fundraising Professionals.

As the Clinic for Special Children's Development Director, Teresa is responsible for providing opportunities for support, stewarding philanthropic relationships, and serving on CSC's leadership team. Prior to joining the Clinic, Teresa was the Associate Director of Development for a continuing care retirement community where she significantly increased revenues for special events and exceeded fundraising goals for the organization. She says of her service in development, "It is rewarding knowing that my work helps provide underserved individuals with state of the art care and essential services that they need and deserve."

KEVIN A. STRAUSS

Medical Director

Kevin Adams Strauss earned a Bachelor of Arts in Biology from Colgate University in 1990. He received his Medical Degree from Harvard Medical School in 1998 and completed his residency in pediatrics at Boston Children's Hospital. In 2001, he joined the Clinic as a Pediatrician. In 2008 he became the Clinic's Medical Director. In this role he has responsibility for the Clinic's clinical and research programs. He is also an Adjunct Associate Research Professor at Franklin & Marshall College. He is a member of the Society of Inherited Metabolic Disorders and the American Academy of Pediatrics. He is board certified in pediatrics by the American Board of Pediatrics.

Dr. Strauss has co-authored over 60 peer reviewed journal articles and has given numerous lectures about medical, scientific, and cultural issues all over the globe. He was awarded the Block Prize for innovation in developmental disabilities research in 2013 and has presented at the Lancaster TEDx event. During residency he received the Michael Osband Memorial Aware for Excellence in Clinical Research, the Medical Student Education Committee Aware for Teaching Excellence, and the Senior Resident Teaching Award.

"We provide a medical home for some of the world's most vulnerable children. These children are vulnerable in every sense of the word"

- Dr. Kevin A. Strauss, Medical Director

Our Services

GENERAL SERVICES

- Amino acid quantification
- Consultation services
- Dietary support
- Genetic counseling and education
- Molecular diagnostic testing
- Routine immunizations
- Molecular cytogenetic analysis
- Well care and sick visits

SPECIALTY MEDICINE

- Audiology
- Behavioral Therapy
- Cardiology
- Endocrinology
- ENT
- Nephrology
- Ophthalmology
- Orthopedics
- Psychology
- Psychiatry
- Pulmonology
- Wheelchair fittings

Supporters & Collaborators

Approximately 90% of our patients are from

THE PLAIN COMMUNITIES

and most do not have health insurance.

We work through a number of third party service providers who graciously donate their services or extend discounts to our patients. We could not provide comprehensive and affordable healthcare without the support of our individual donors, collaborators and supporters.

National Press Coverage

The New York Times

THE BALTIMORE SUN

THE WALL STREET JOURNAL.

NBC NEWS

USA TODAY

The Philadelphia Inquirer

The Washington Post

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